



autosomal recessive primary microcephaly

Autosomal recessive primary microcephaly (often shortened to MCPH, which stands for "microcephaly primary hereditary") is a condition in which infants are born with a very small head and a small brain. The term "microcephaly" comes from the Greek words for "small head."

Infants with MCPH have an unusually small head circumference compared to other infants of the same sex and age. Head circumference is the distance around the widest part of the head, measured by placing a measuring tape above the eyebrows and ears and around the back of the head. Affected infants' brain volume is also smaller than usual, although they usually do not have any major abnormalities in the structure of the brain. The head and brain grow throughout childhood and adolescence, but they continue to be much smaller than normal.

MCPH causes intellectual disability, which is typically mild to moderate and does not become more severe with age. Most affected individuals have delayed speech and language skills. Motor skills, such as sitting, standing, and walking, may also be mildly delayed.

People with MCPH usually have few or no other features associated with the condition. Some have a narrow, sloping forehead; mild seizures; problems with attention or behavior; or short stature compared to others in their family. The condition typically does not affect any other major organ systems or cause other health problems.

Frequency

The prevalence of all forms of microcephaly that are present from birth (primary microcephaly) ranges from 1 in 30,000 to 1 in 250,000 newborns worldwide. About 200 families with MCPH have been reported in the medical literature. This condition is more common in several specific populations, such as in northern Pakistan, where it affects an estimated 1 in 10,000 newborns.

Genetic Changes

MCPH can result from mutations in at least seven genes. Mutations in the *ASPM* gene are the most common cause of the disorder, accounting for about half of all cases.

The genes associated with MCPH play important roles in early brain development, particularly in determining brain size. Studies suggest that the proteins produced from many of these genes help regulate cell division in the developing brain.

Mutations in any of the genes associated with MCPH impair early brain development. As a result, affected infants have fewer nerve cells (neurons) than normal and are born

with an unusually small brain. The reduced brain size underlies the small head size, intellectual disability, and developmental delays seen in many affected individuals.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- MCPH
- microcephaly primary hereditary
- primary autosomal recessive microcephaly
- true microcephaly

Diagnosis & Management

These resources address the diagnosis or management of MCPH:

- GeneReview: Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders
<https://www.ncbi.nlm.nih.gov/books/NBK9587>
- Genetic Testing Registry: Primary autosomal recessive microcephaly 1
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1855081/>
- Genetic Testing Registry: Primary autosomal recessive microcephaly 2
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858535/>
- Genetic Testing Registry: Primary autosomal recessive microcephaly 3
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858108/>
- Genetic Testing Registry: Primary autosomal recessive microcephaly 4
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858516/>
- Genetic Testing Registry: Primary autosomal recessive microcephaly 5
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1837501/>
- Genetic Testing Registry: Primary autosomal recessive microcephaly 6
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842109/>
- Genetic Testing Registry: Primary autosomal recessive microcephaly 7
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2675187/>
- MedlinePlus Encyclopedia: Head Circumference
<https://medlineplus.gov/ency/article/002379.htm>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Head Circumference
<https://medlineplus.gov/ency/article/002379.htm>
- Encyclopedia: Microcephaly
<https://medlineplus.gov/ency/article/003272.htm>
- Encyclopedia: Microcephaly (image)
<https://medlineplus.gov/ency/imagepages/17256.htm>
- Health Topic: Brain Malformations
<https://medlineplus.gov/brainmalformations.html>

Genetic and Rare Diseases Information Center

- Autosomal recessive primary microcephaly
<https://rarediseases.info.nih.gov/diseases/12117/autosomal-recessive-primary-microcephaly>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Microcephaly
<https://www.ninds.nih.gov/Disorders/All-Disorders/Microcephaly-Information-Page>

Educational Resources

- Boston Children's Hospital
<http://www.childrenshospital.org/conditions-and-treatments/conditions/microcephaly>
- Centers for Disease Control and Prevention: Intellectual Disability
https://www.cdc.gov/ncbdd/actearly/pdf/parents_pdfs/IntellectualDisability.pdf
- Cleveland Clinic
<http://my.clevelandclinic.org/childrens-hospital/health-info/diseases-conditions/hic-Microcephaly>
- Disease InfoSearch: Primary autosomal recessive microcephaly 1
<http://www.diseaseinfosearch.org/Primary+autosomal+recessive+microcephaly+1/9155>
- Disease InfoSearch: Primary autosomal recessive microcephaly 2
<http://www.diseaseinfosearch.org/Primary+autosomal+recessive+microcephaly+2/9156>
- Disease InfoSearch: Primary autosomal recessive microcephaly 3
<http://www.diseaseinfosearch.org/Primary+autosomal+recessive+microcephaly+3/9157>
- Disease InfoSearch: Primary autosomal recessive microcephaly 4
<http://www.diseaseinfosearch.org/Primary+autosomal+recessive+microcephaly+4/9158>
- Disease InfoSearch: Primary autosomal recessive microcephaly 5
<http://www.diseaseinfosearch.org/Primary+autosomal+recessive+microcephaly+5/9159>
- Disease InfoSearch: Primary autosomal recessive microcephaly 6
<http://www.diseaseinfosearch.org/Primary+autosomal+recessive+microcephaly+6/9160>
- Disease InfoSearch: Primary autosomal recessive microcephaly 7
<http://www.diseaseinfosearch.org/Primary+autosomal+recessive+microcephaly+7/9161>
- Lucille Packard Children's Hospital
<http://www.stanfordchildrens.org/en/topic/default?id=microcephaly-90-P02610>

- MalaCards: primary autosomal recessive microcephalies and seckel syndrome spectrum disorders
http://www.malacards.org/card/primary_autosomal_recessive_microcephalies_and_seckel_syndrome_spectrum_disorders
- Orphanet: Autosomal recessive primary microcephaly
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2512

Patient Support and Advocacy Resources

- Resource list from the University of Kansas Medical Center: Brain (Cephalic) Conditions
<http://www.kumc.edu/gec/support/brain.html>

GeneReviews

- Primary Autosomal Recessive Microcephalies and Seckel Syndrome Spectrum Disorders
<https://www.ncbi.nlm.nih.gov/books/NBK9587>

Genetic Testing Registry

- Primary autosomal recessive microcephaly 1
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1855081/>
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<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1842109/>
- Primary autosomal recessive microcephaly 7
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2675187/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22primary+autosomal+recessive+microcephaly%22+OR+%22Microcephaly%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Microcephaly%5BMAJR%5D%29+AND+%28%28primary+autosomal+recessive+microcephaly%5BTIAB%5D%29+OR+%28mcph%5BTIAB%5D%29+OR+%28primary+microcephaly%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- MICROCEPHALY 1, PRIMARY, AUTOSOMAL RECESSIVE
<http://omim.org/entry/251200>
- MICROCEPHALY 2, PRIMARY, AUTOSOMAL RECESSIVE, WITH OR WITHOUT CORTICAL MALFORMATIONS
<http://omim.org/entry/604317>
- MICROCEPHALY 3, PRIMARY, AUTOSOMAL RECESSIVE
<http://omim.org/entry/604804>
- MICROCEPHALY 4, PRIMARY, AUTOSOMAL RECESSIVE
<http://omim.org/entry/604321>
- MICROCEPHALY 5, PRIMARY, AUTOSOMAL RECESSIVE
<http://omim.org/entry/608716>
- MICROCEPHALY 6, PRIMARY, AUTOSOMAL RECESSIVE
<http://omim.org/entry/608393>
- MICROCEPHALY 7, PRIMARY, AUTOSOMAL RECESSIVE
<http://omim.org/entry/612703>

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